SACGT Report To CLIAC

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SACGT Focus: White Paper

- Initial White Paper outlined key features of genetic testing, emphasizing need to improve test availability, quality, oversight functions
- Concerns included:
  - Definition of genetic test
  - How to improve quality but retain access
  - Knowledge base about current tests and when and how to use them
  - Patient protection for sensitive tests
  - Others

SACGT Focus: White Paper

- Some Areas of Overlap Interest with CLIAC:
  - Definition of a genetic test: CLIAC definition (heritable and acquired disease)
  - Charged FDA to develop ability for pre-market review of genetic tests
  - Charged CDC, CMS to advance expanded laboratory oversight

SACGT Work Groups

- Data Work Group
- Education Work Group
- IRB/Consent Work Group*
- Rare Diseases Work Group*
- Access Work Group*

Data Work Group

- **Goal: To improve knowledge of diseases clinical utility of tests (predictive value**
- **Need to improve data collection, and analysis**
- **Both clinical and laboratory data required**

Lab Issues:

- Definition of a test: Confusion between "disease" and "test

-Who is to provide the data and how? Privacy? Cost? Structure and analysis?

Education Work Group

- **Need: Learn activities in current genetics education, results of a roundtable meeting held 11/14 were reported. Key elements included**
  - consideration of groups that require education,
  - need for an evidence-based approach
  - need to define core competencies
- **Workshop is planned for May**

Consent/IRB Work Group

- **Brochure was developed to serve as a model of explaining genetic testing and informed consent to the general public. It was presented and reviewed**
- **White paper is under development on principles of informed consent, including issues pertaining to determination of the level of consent required for different kinds of genetic tests**

<u>Issues:</u> Who decides what type of consent is needed? FDA role? Role of professional societies? Decisions by disease or by test? Laboratory's role and responsibility?

Access Work Group

Focus:

- Patient need for information
- Reimbursement issues
  - Views of payors
  - Test cost, (Non-reimbursed laboratory costs)
  - Counseling needs and costs
- Patents (access and quality)

Rare Disease Testing Work Group

- **Dr. Whittemore: (Tuberosclerosis model) described approach to CLIA approved laboratory testing of a rare disease.**
- **Dr. Haffner, Dir. FDA Office of Orphan Products described FDA approach to supporting products for orphan diseases**
- **Dr. Less, Director of Investigational Device Exemption (IDE) and Humanitrian Device Exemption (HDE) described processes for limited usage products.**

Rare Disease Testing Work Group

- Dr. Hyatt-Knorr, Acting Director of NIH Office of Rare Diseases (ORD) described the office
- Dr. Miller (Genzyme) discussed commercial development of genetic tests, issues of informed consent, knowledge base of disease etc.
- Dr. Wenger (Jefferson), and Dr. Ledbetter, (U. Chicago) discussed approaches to offer orphan lab tests through partnerships with CLIA labs

Rare Disease Testing Work Group

Goal:

- To develop knowledge of when and how to test for rare diseases
- To develop access to quality testing for rare diseases
  - Current testing is often performed in research laboratories that are uneven
  - Those doing the testing are not properly supported for this work in education, funding, resources
- A White Paper is being developed

Pre-market Approval of Patient Care Tests

- **Strategy:**

  FDA review of all new genetic tests ready for "prime time" patient care

- **Concerns:**

  a) **The number of tests exceeds FDA capacity**

  b) **Regulatory restrictions cannot be burdensome**

Test Review Template

- **Test name, intended use, indications**
- **Methodology, procedure manual**
- **Provide examples of tests**
- **Documentation of analytic validity**
  (includes tested, results, sensitivity, specificity)
- **Quality control procedures**
- **Documentation of clinical validity**
- **Clinical interpretation, test limitations,**
- **Clinical utility if known**

CLIAC Report to SACGT

- Waived tests
- CMS study of laboratories performing waived tests

Request of Chair (Dr. Mc Cabe)

- Request from the HHS agencies information regarding their efforts to advance knowledge of genetic tests in four core activities:
  - Primary research
  - Secondary analysis of existing data
  - Projects for developing or updating knowledge
  - Information dissemination projects for public, others